

Note: The following information is provided by the author(s) and has not been reviewed by GeneReviews staff.

Table 2. Point Mutations Detected in Individuals with FRDA Who Are Compound Heterozygous

Exon/Intron	Mutation	Nt Position	NT Change	Predicted Effect on Frataxin	Reference
Exon 1	M1L	1	ATG>CTG	Aberrant initiation of translation	[Cossee et al 1999]
Exon 1	M1T	2	ATG>ACG	Aberrant initiation of translation	[Cossee et al 1999]
Exon 1	2delT	2	ATG>A-G	Aberrant initiation of translation	[Zhu et al 2002; Bidichandani et al, unpublished]
Exon 1	M1I	3	ATG>ATT	Aberrant initiation of translation	[Cossee et al 1997]
Exon 1	M1I	3	ATG>ATA	Aberrant initiation of translation	[Potter et al 2000]
Exon 1	11del2	11	del TC	Premature termination of translation	[Spacey et al 2004]
Exon 1	100delG	100	del G	Premature termination of translation	[Gellera et al 1997]
Exon 1	104delC	104	del C	Premature termination of translation	[Gellera et al 1997]
Exon 1	R40C	118	CGT>TGT	Missense	[Van Driest et al 2005]
Exon 1	118delC	118	del C	Premature termination of translation	[De Castro et al 2000]
Exon 1	158delC	158	del C	Premature termination of translation	[Cossee et al 1999]
Exon 1	158insC	158	ins C	Premature termination of translation	[Cossee et al 1999]
Intron 1	Splice donor	+5	G>C	Aberrant splicing	[McCormack et al 2000]
Exon 2	202GTCA→TTG	202-205	GTCA>TTG	Premature termination of translation	[Pook et al 2000]
Exon 3	297insT	297	ins T	Premature termination of translation	[De Castro et al 2000]
Exon 3	L106X	317	TTA>TGA	Premature	[Campuzano et al

Exon/Intron	Mutation	Nt Position	NT Change	Predicted Effect on Frataxin	Reference
				termination of translation	[1996]
Exon 3	L106S	317	TTA>TCA	Missense	[Bartolo et al 1998]
Exon 3	317delT	317	del T	Premature termination of translation	[Pook et al 2000]
Exon 3	340del13	340-352	del of 13 bp	Premature termination of translation	[Pook et al 2000]
Exon 3	Y118X	354	TAC>TAG	Premature termination of translation	[Gellera et al 1997]
Exon 3	D122Y	364	GAC>TAC	Missense	[Gellera et al 1997]
Exon3/Intron3	381_384 delTGGG+g.IVS3+1_+9	381	del of 11689 bp	Aberrant splicing	[Gellera et al 1997]
Intron 3	Splice donor	384+1	AG>GG	Aberrant splicing	[Doudney et al 1997]
Intron 3	Splice acceptor	-2	AG>GG	Frameshift due to aberrant splicing of exon 4	[Campuzano et al 1996]
Exon 4	G130V ¹	389	G130V 1	Missense; aberrant processing	[Bidichandani et al 1997, Koutnikova et al 1998]
Exon 4	N146K	438	C>G	Missense	[Zühlke et al 2004]
Exon 4	Q148R	443	CAG>CGG	Missense	[McDaniel et al 2003]
Exon 4	I154F ¹	460	ATC>TTC	Missense; aberrant processing	[Campuzano et al 1996, Koutnikova et al 1998]
Exon 4	W155R	464	TGG>CGG	Missense	[Labuda et al 2000]
Exon 4	W155X	465	TGG>TAG	Premature termination of translation	[De Castro et al 2000]
Exon 4	L156P	467	CTA>CCA	Missense	[Cossee et al 1999]
Intron 4-exon 5a	g.120032_1222808del	g.120032	del of 2776 bp	Deletion of exon 5a	[Zühlke et al 2004]
Intron 4	Splice donor	482+2	GT>GG	Frameshift due to aberrant splicing of exon 4	[Forrest et al 1998]
Intron 4	Splice donor	482+3	+3 del A	Frameshift due to aberrant splicing of exon 4	[Cossee et al 1999]
Exon 5a	R165C ¹	493	CGT>TGT	Missense	[Forrest et al 1998, McCormack et al

Exon/Intron	Mutation	Nt Position	NT Change	Predicted Effect on Frataxin	Reference
					2000]
Exon 5a	R165P	494	CGT>CCT	Missense	[De Michele et al 2000]
Exon 5a	W173G ¹	517	TGG>GGG	Missense	[Cossee et al 1999]
Exon 5a	L182F	544	CTC>TTC	Missense	[Forrest et al 1998]
Exon 5a	L182H	545	CTC>CAC	Missense	[Cossee et al 1999]
Exon 5a	H183R	548	CAT>CGT	Missense	[Cossee et al 1999]
Exon 5a	L186R	557	T>G	Missense	[Zühlke et al 2004]
Exon 5a	L198R	593	CTG>CGG	Missense	[Al-Mahdawi et al 2000]

1. Recurrent mutations

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